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Letter to the Editor

A single case report lacking details does not equal a mimic of abusive head trauma

Dear Editor,

We read with interest the recent case report by De Leeuw et al. regarding delta-storage pool disease; however, we disagree with the conclusion that delta-storage pool disease should be considered a "mimic" of abusive head trauma (AHT).[1]

The clinical and radiographic findings and clinical outcomes associated with AHT have been well characterized. Multiple SDH over the convexity, interhemispheric hemorrhages often accompanied by apnea, seizures, rib or other fractures and head/neck bruising are often seen, along with poor neurological outcomes.[2–5] These associations are statistically significant over the breadth of the medical literature. Additionally, the retinal hemorrhages often seen in AHT are of greater severity that those seen in other conditions.[6] These are specific, detailed findings. To be a "mimic" of AHT, another condition should 1) cause the same or very similar detailed findings that are seen in true AHT, and 2) cause these findings in a large number of cases, or, better yet, be statistically associated with the presence of these specific findings. In claiming that delta-storage pool disease is a "mimic" of AHT, De Leeuw et al. present a single case report lacking critical specific details of a child diagnosed with a mild platelet disorder. The authors attribute the child's findings to the mild platelet disorder without reasonably considering that the child may have been a victim of AHT, as having a mild platelet disorder does not protect one from AHT.

Literature characterizing bleeding symptoms specifically in delta-storage pool disease is lacking. Bleeding manifestations of platelet storage pool disorders as a whole are usually mild. Individuals present with mucocutaneous bleeding or "easy bruising".[7–10] Excessive bleeding from the umbilical stump, after circumcision or vaccinations, or while teething has been noted. Prolonged epistaxis is a common presenting complaint, as are menorrhagia and post-partum bleeding. Significant bleeding, such as hemarthroses and muscle hematomas are rare. Many individuals do not present until later in life, after a hemostatic challenge. In summary, the existent literature indicates that bleeding symptoms in delta-storage pool disease and other platelet storage pool disorders is most often mild in nature.

The main question raised by this case report is: can delta-storage pool disease result in subdural hematoma (SDH), retinal hemorrhages (Rh), brain edema and long term brain damage in the setting of minimal or no trauma? This case report does not provide useful information regarding this question for several reasons. First, it is possible to have both delta-storage pool disease and AHT. This case report lacks sufficient information to reasonably determine that AHT had been completely evaluated: no information on initial or repeat skeletal surveys, detailed descriptions of retinal hemorrhages, or images of MRI or CT scans was provided, and details of the initial clinical presentation regarding apnea and/or seizures are missing. Additionally, even if one accepts that delta-storage pool disease can cause SDH and Rh, no physiologic rationale for the involved child's persistent brain damage was presented. Much of the authors' conclusions regarding the role of delta-storage pool disease in the case report appear to be based on rebleeding of the initial SDH, which is a known potential complication of any SDH, regardless of inciting cause.[11] Lastly, this case report stands in stark contrast to the known presentations of delta-storage pool disease and other mild platelet function disorders: mucocutaneous bleeding, "easy bruising" and bleeding in response to hemostatic challenges.

Using a single case report that lacks critical details to guide forensic diagnosis is scientifically unacceptable. Case reports should inform testable hypotheses, such as, "delta-storage pool disease may cause SDH, Rh, cerebral edema, and poor neurological outcomes in the presence of minimal or no trauma." To test this hypothesis, patients with delta-storage pool disease should be compared to those without a bleeding disorder for the presence of the outcomes of interest. No such studies exist; thus, asserting that delta-storage pool disease is a "mimic" of AHT is scientifically inaccurate, particularly in the context of significant literature showing the association and predictive values of the outcomes of interest with AHT.[2–6]

Conflict of interest

None declared.

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James Anderst, MD, MSCI, Associate Professor*
Division of Child Abuse and Neglect, Children's Mercy Hospital, University of Missouri Kansas City School of Medicine, Kansas City, MO, USA

Shannon Carpenter, MD, MSCI, Associate Professor
Division of Hematology/Oncology, Children's Mercy Hospital, University of Missouri Kansas City School of Medicine, Kansas City, MO, USA
E-mail address: slcarpenter@cmh.edu (S. Carpenter)

* Corresponding author.
E-mail address: jdanderst@cmh.edu (J. Anderst)

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